A case of Diastrophic Dysplasia
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Objective
Herein we present a case of diastrophic dysplasia, with the aim to illustrate prenatal sonographic findings and postnatal correlation to the disease.

Methods
This is a case report.

Results
21 year old P0G0 woman, without consanguinity or family history of any genetic disorders, was referred to Cairo University Fetal Medicine Unit, because of suspicion of lethal skeletal dysplasia, raised during a scan one week prior to our scan. Our ultrasound examination revealed the following findings; polyhydramnios, micromelia, non-lethal thoracic hypoplasia, micrognathia, talipes, hitchhiker thumb, malformed external ear and square face. There was absence of cleft palate. Based on our ultrasound examination we reached the diagnosis of diastrophic dysplasia, which was also confirmed postnatally.

Conclusion
Homozygous mutation in the DTDST gene (diastrophic dysplasia sulfate transporter gene) results in diastrophic dysplasia, which is inherited in an autosomal recessive pattern. Mutation in this gene results in a spectrum of skeletal dysplasias, which are variable in severity and consist of Achondrogenesis Type 1, Atelosteogenesis Type 2 and recessive multiple Epiphyseal Dysplasia, in addition to Diastrophic Dysplasia. The ultrasound features of diastrophic dysplasia are characteristic and include micromelia, often with curvature, hitchhiker thumb (proximal insertion with fixed abduction), which is considered as the pathognomonic feature of diastrophic dysplasia, and other digital malformations. Other common findings include limitation in mobility of the joints, talipes, scoliosis, abnormal "cauliflower" ears and cleft palate in some occasions. The highest incidence of this rare skeletal dysplasia is present among Finns. Diastrophic dysplasia is not lethal. However, neonatal mortality rate is estimated to be about 25%, mainly due to laryngotracheomalacia and stenosis. Volumetric ultrasound technique is superior to usual two-dimensional studies in depicting limb anomalies and specifically facial dysmorphism.